# SLC52A2 gene

solute carrier family 52 member 2

## Normal Function

The *SLC52A2* gene provides instructions for making a riboflavin transporter protein called RFVT2 (formerly known as RFT3). This protein moves (transports) a vitamin called riboflavin (also called vitamin $B_2$) across the cell membrane. The RFVT2 protein is found at especially high levels in cells of the brain and spinal cord and is important for absorbing riboflavin from the bloodstream into these tissues.

In the cells of the body, including those in the brain and spinal cord, riboflavin is the core component of molecules called flavin adenine dinucleotide (FAD) and flavin mononucleotide (FMN). These molecules function as coenzymes, which means they help enzymes carry out chemical reactions. FAD and FMN are involved in many different chemical reactions and are required for a variety of cellular processes. One important role of these coenzymes is in the production of energy for cells. FAD and FMN are also involved in the breakdown (metabolism) of carbohydrates, fats, and proteins.

## Health Conditions Related to Genetic Changes

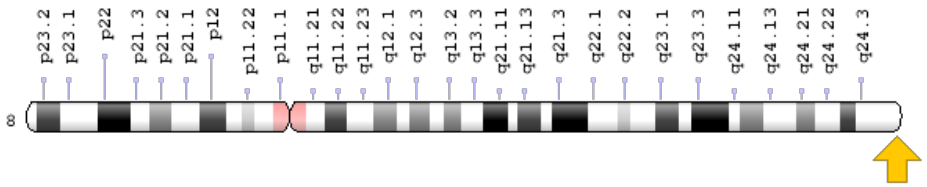
riboflavin transporter deficiency neuronopathy

At least 11 mutations in the *SLC52A2* gene have been found to cause riboflavin transporter deficiency neuronopathy. This neurological condition encompasses two disorders that were previously considered to be separate: Brown-Vialetto-Van Laere syndrome and Fazio-Londe disease. Some of the gene mutations involved in riboflavin transporter deficiency neuronopathy prevent production of the RFVT2 protein. Others lead to production of an abnormal protein with impaired ability to transport riboflavin. It is unclear how these changes lead to the nerve problems that cause hearing loss, muscle weakness in the face and limbs, and breathing problems in people with the disorder.

## Chromosomal Location

Cytogenetic Location: 8q24.3, which is the long (q) arm of chromosome 8 at position 24.3

Molecular Location: base pairs 144,358,547 to 144,361,286 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- BVVLS2
- D15Ertd747e
- FLJ11856
- G protein-coupled receptor 172A
- GPCR41
- GPR172A
- hRFT3
- PAR1
- PERV-A receptor 1
- porcine endogenous retrovirus A receptor 1
- putative G-protein coupled receptor GPCR41
- RFT3
- RFVT2
- riboflavin transporter 3
- solute carrier family 52 (riboflavin transporter), member 2
- solute carrier family 52, riboflavin transporter, member 2

## Additional Information & Resources

GeneReviews

- Riboflavin Transporter Deficiency Neuronopathy
  https://www.ncbi.nlm.nih.gov/books/NBK299312

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SLC52A2%5BTIAB%5D%29+OR+%28solute+carrier+family+52++,+member+2%5BTIAB%5D%29%29+OR+%28%28G+protein-coupled+receptor+172A%5BTIAB%5D%29+OR+%28GPCR41%5BTIAB%5D%29+OR+%28GPR172A%5BTIAB%5D%29+OR+%28RFT3%5BTIAB%5D%29+OR+%28RFVT2%5BTIAB%5D%29+OR+%28hRFT3%5BTIAB%5D%29+OR+%28putative+G-protein+coupled+receptor+GPCR41%5BTIAB%5D%29+OR+%28riboflavin+transporter+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 52 (RIBOFLAVIN TRANSPORTER), MEMBER 2
  http://omim.org/entry/607882

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC52A2%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=30224
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/79581
- UniProt
  http://www.uniprot.org/uniprot/Q9HAB3

## Sources for This Summary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